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ABSTRACT

Presented are two papers from a panel discussion on prenatal diagnosis and genetic counseling with families. D. Blackston (director of the Developmental Evaluation Clinic, Decatur, Georgia) points out that a concise family history, pregnancy and birth data, developmental history, careful physical examination, and appropriate laboratory studies are essential to effectively deal with handicapping conditions and to properly inform parents. In the second paper, I. L'Abate (professor of psychology and director of the Family Studies Program at Georgia State University, Atlanta) discusses the need for sharing feelings when helping families deal with hurt and pain. (SBH)

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INTRODUCTION: CEC

Family Assessment & Genetic Counseling

Pat Carpenter

Working with visually impaired and multihandicapped children and their parents gives me a very strong desire to prevent congenital anomalies and birth defects before they happen. Recent developments in prenatal diagnosis and genetics has increased knowledge of hereditary conditions and the genetic traits which affect live births.

The first step for a family needing genetic counseling is to recognize the need. This may come about through personal concern about recurrence of a condition within the family, or may be suggested by a doctor.

Family data must be collected and graphed in the form of a pedigree for analysis to determine a pattern of inheritance.

When genetic studies are undertaken, history, past medical history, direct physical examination and laboratory evaluation of individuals within the pedigree.

Once genetic studies are completed in a given family, the genetic counselor will seldom prescribe a particular course of action. The counselor's role is to provide the family with a realistic view of the situation - the nature of the birth defect already manifested in a family member or members, the risk of recurrence and what recurrence would mean in practical terms of all concerned.

The members of our panel are well qualified to discuss some of the defects which are a result of genetic inheritance, and the interpersonal dynamics in the family constellation which impinge upon the decision-making process for families under stress.

Dr. Dwain Blackston, Director of the Developmental Evaluation Clinic, DeKalb Health Department, is a pediatrician who examines many babies with birth defects, counsels families, and evaluates children's potential for living and learning. He works closely with us in special education.

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Dr. Luciano L'Abate, Director of Family Studies at Georgia State University, is a psychologist who has developed a battery of materials for assessment of family strengths and weaknesses and the roles that various family members take in relation to each other. He and his wife together do family counseling.

Dr. Zane Pollard, Pediatric Ophthalmologist, is very concerned about the genetic inheritance of visual defects. I particularly like to receive one of his eye reports because they always include genetic factors as they relate to a child's vision.

COUNCIL FOR EXCEPTIONAL CHILDREN
FAMILY ASSESSMENT AND GENETIC COUNSELLING

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In order to deal with handicapping conditions and birth defects, one must have a basic understanding of the essential data base to impart precise diagnosis, counselling and to offer some prognosis as to the outlook for the handicapped child. Today we want to pay particular attention to that data base which includes careful family history or pedigree; careful pregnancy, delivery, and developmental history; and very careful physical examination of the handicapped child. Further testing, such as chromosome analysis, metabolic screening for enzyme disorders, and other helpful studies such as x-rays are ordered on the basis of the history and physical examination. The history and physical give one the very basic concepts as to whether or not the situation is a single gene defect, either autosomal dominant or recessive, whether or not there are multiple congenital defects suggestive of chromosomal disorders, or whether there are findings highly suggestive of a metabolic or enzymatic disorder, with most of these being primarily recessive disorders. One must be careful to include in history taking a careful assessment as to whether or not there was any prenatal infection such as rubella or toxoplasmosis or whether there was any history of teratogens such as medications or chronic alcohol intake. The following slides will illustrate this proposed methodology as well as show classic examples of autosomal dominant inheritance, autosomal recessive inheritance, multi-factorial inheritance, chromosomal disorders, prenatal infection, and teratogenic disorders.

Single gene disorders are classified as either autosomal dominant, autosomal recessive, or sex linked. They may be very mild or they may be quite severe with multi-system involvement. In particular, eye involvement may be none to very severe. An autosomal dominant inheritance pattern is demonstrated showing that autosomal dominant affects either sex equally, and if a parent has an autosomal dominant trait,

he can transmit this to his children at a 50% recurrence risk for every child. In general, dominant traits are not so severe as the recessive ones, and they are also characterized by a fairly high mutation rate and low penetrance rate. Careful history and physical examination can elucidate the mild forms or low penetrance forms and can be helpful in genetic counseling. The following family demonstrates an autosomal dominant trait transmitted from the mother to 3 of her 4 children, whereby they had microphthalmia or small eyes, and central cataracts resulting in visual impairment. This is a well-informed, well-adjusted family seeking genetic counseling primarily because the oldest daughter was married and wished children of her own. She also wanted to know at what risk she would be for having children with difficulties similar to her own. Since this clearly showed as a dominant trait, she was counseled that for each pregnancy she would be at a 50% or 1 in 2 risk for her children to have the same disorder. With this information carefully and sensitively disseminated to this young woman, she and her husband decided they wanted children of their own and decided to take the risk of having a handicapped child. They did, indeed, have a child with visual problems very similar to those of the mother. They are happy, well-adjusted parents who are coping well and who can anticipate well the needs of this child with possible visual difficulties. It must be stressed at this point that the role of the genetic counselor is to inform and understand that the ultimate decision for children and for taking genetic risk lies privately and personally with the parents. This is, indeed, not a case of failure of genetic counseling, but an example of excellent genetic counseling and decision made on the basis of knowing what the problems can be and on having effectively dealt with them on a personal basis by this mother and her family.

Autosomal recessive disorders are those in which both parents must be carriers and that these two abnormal genes or alleles result in full-blown recessive disorder. As depicted, recessive disorders can affect either sex equally and parents who are carriers of a recessive disorder have a 25% risk for each pregnancy result-

ing in the carried disorder. Furthermore, half their children will be at risk for being carriers. An example of a recessive disorder is the Laurence-Moon-Biedl Syndrome shown in this short, obese, mentally retarded, visually-impaired girl whose eye examination did show retinitis pigmentosa with the latter finding as well as the aforementioned ones adding up to the cardinal signs or findings of the Laurence-Moon-Biedl Syndrome. Clinically, these people begin to get visual difficulties early in life with the first symptoms being night blindness. A great majority are severely visually impaired or blind by age 20. The exact cause of the Laurence-Moon-Biedl Syndrome is unknown.

Chromosomal aberrations, be they extra material, so called trisomy, or deleted or missing material, may result in severe multisystem involvement. The involvement of the eye is only a part of the total effect of these chromosome aberrations. Chromosomal abnormalities almost always have also rather marked developmental delays, particularly if they involve the autosomes; whereas, sex chromosome abnormalities may have little to no physical or developmental abnormalities. Chromosomal abnormalities do not follow Mendelian patterns of inheritance such as dominant or recessive but are the result of chromosome misdivision, usually very early in the meiosis of the germ cells. In all cases where there is a previous chromosomal disorder we offer prenatal monitoring or amniocentesis to the parents. This, again, is a personal, private decision based upon recurrence risks which generally are low, mainly in the neighborhood of 1 to 2%, and with the indication that therapeutic abortion may be necessary should chromosomal aberrations be found. While it is unlikely that parents would not want therapeutic abortion should a chromosomal abnormality be found, again, this is a personal and private decision for those parents. It is imperative that both parents be involved in the informing and counseling sessions at all times. The first chromosomal aberration I will show to you is the so called Cat-Eye Syndrome in an infant who had abnormal ears, preauricular tags, imperforate anus, congenital heart disease, and coloboma

of the iris. The preauricular tags and the abnormal ears are easily seen in the first slide, while the second slide shows the colobomata of the iris resulting in eyes that look very much like cat eyes. This is the result of a small, extra acrocentric chromosome believed to be chromosome number 22 as shown. The next slide depicts for you a case of Trisomy 13, showing a child with severe cleft lip and palate, severe micropthalmia and abnormal ears. Most of these children also have severe involvement of the heart, kidneys and development of the forebrain; and, the prognosis for life is quite poor. The next picture shows classic epicanthal folds and Brushfield spots, which are commonly seen in children with Down's Syndrome, the most common of the autosomal defects, occurring in 1:600 live births. Down Syndrome children are at a much higher risk for cataracts and other eye abnormalities also.

Multi-factorial inheritance is the geneticist's nightmare because it is often difficult to give precise counseling in these situations, and, at times, virtually impossible to rule out recessive inheritance patterns. Multi-factorial inheritance means the effects of several abnormal genes acting in unison with environmental influences resulting in single or multiple congenital birth defects. Classic examples of multi-factorial inheritance are cleft lip and palate and neural tube defects which occur at about a 5% recurrence rate for a second child to have this, if parents have had a previous child with these defects. The example given to you is that of a child with holoprosencephaly syndrome, showing severe cleft lip and palate, severe microphthalmia and a sloping forehead with poor development of the forebrain. This series of findings is felt to represent a sporadic event and multi-factorial risks for recurrence are given to parents. This obviously is also based heavily upon family data studies and other statistics with the always nagging problem that the recessive mode cannot be fully ruled out.

Congenital birth defects, in particular eye defects, can be the results of prenatal infections or teratogens, agents which result in abnormal structure or morphology of the fetus. The classic example of prenatal infection is that

of Congenital Rubella Syndrome and the slides show a child with congenital glaucoma as well as a child with bilateral cataracts. Additionally, children with congenital rubella may have hearing impairments, congenital heart disease, and other difficulties. A recently well-documented teratogen is alcohol, and the following slides illustrate to you a child whose mother was a chronic alcoholic with the child having low birth weight, very small eyes, and cleft palate. It is projected that the Fetal Alcohol Syndrome is, indeed, a public health problem with approximately 0.8:1000 mothers having high alcohol intake, and it is also the projection that some 30 to 40% of their offspring will be low in birth weight and will have delayed growth and development. One, thus, has to give consideration to remediation for this condition before becoming pregnant, and possibly at the therapeutic abortion could be offered to these people who are at very high risk. Alcohol is believed to have a toxic effect upon the developing fetus, and it is felt that it is not the nutritional deficiencies imposed by chronic alcoholism that result in the impaired infant.

In summary, to effectively deal with handicapping conditions and to properly inform parents a concise family history, pregnancy and birth history, developmental history, careful physical examination and appropriate laboratory studies for objective documentation of the diagnosis are essential. It must be pointed out that this can be done with confidence, sensitivity and with imparting of great reassurance to parents. The role of genetic counselor is an intimate one with parents, but it must be emphasized that of an informed, supportive counselor rather than advisor. Decisions as to intervention, decision as to further children, and decision as to whether or not to have prenatal monitoring are ultimate and private decisions of the parents.

Family Assessment and Genetic Counseling

## Helping Families Deal With Hurt and Pain

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How do we deal with hurt? How do we deal with a hurt system? I do want to spend some time about talking with the family that receives this kind of painful information or is faced by this kind of painful event, trauma, or disease. We are dealing essentially with a hurt family, and I do believe that we are, culturally and socially, untrained to deal with hurt. And I am not only speaking for people, I am talking also and especially about professionals. We do not know how to deal with our own hurts. If we do not know how to deal with hurt ourselves, how can we help others? Clearly, it is easy to say, "Support the system." It is not easy to tell them, to evaluate them, to make them understand why something of this proportion took place. It is even harder to deal with the aftermath, with the ripples of the crisis, with the conflicts that derive from it, with the stress on the system. Sometimes the pain and hurt may not be readily apparent. It may influence the family for years to come. What each of us needs to be sensitive to is the grieving, the mourning, and the hurt that is taking place in the family system.

We also need to pay attention, very closely, to sex differences on how we deal with hurt. There are going to be extreme reactions, ranging from passive apathy to anger, acting out, blaming, and denial. The best way to deal with these is to focus on the pain and the hurt. Now, some people will tell you that we should not. Some will say that we should not open that can of worms. I am completely against that position. I do believe that we need to help the

family to share the hurt constructively. And if we don't do it, nobody else will and the lack of expression of hurt feelings may destroy that family.

That means that we have to pay attention to crying, and not be afraid of somebody crying. We need to reinforce and encourage whoever in the family is crying, pointing out that that person who is crying is the lucky one in the family, because this can be congruent in the expression of pain. It is unfortunate for those in the family who cannot cry, who cannot express congruently the pain and hurt that comes from something like this happening. Stereotypically it is easier for women to cry and to express hurt. I suspect that women live longer because they can cry! The problem is the father who is frightened to death by his wife's hurt and tears, and by the other children's crying.

If there is any taboo in our culture, I submit to you that taboo is crying. We do not know how to deal with it. The evidence that we have gathered is based on battered wives, battered children, and raped women. The stimulus that provokes the attack most often is crying. Most of us men are completely inadequate in dealing with this particular expression of feeling. We deny our vulnerability as human beings. We are dealing, therefore, with a system that, in most cases, will not accept fully the expression of hurt. Part of the very same system will deny it. You will find the man who becomes reasonable, who tries to assuage his wife's tears by being reasonable, by being logical, or by going impassively into action, by having to find "band aids" or finding operations or some kind of panacea that, somehow, is magically going to lower the level of hurt in the family. Let us not fall into that trap.

There is absolutely nothing to do with hurt but share it. I'd like to make this point clear. There is nothing to do with hurt in a family but share it. This in itself is an important goal to reach because it is a very difficult one. The grieving has to be dealt with, the mourning has to be expressed whenever there is a loss. When you see a hurt child, a maimed child, a child

who cannot see, there is a loss. Given this particular loss, how can we help this family sum up the assets? That sounds crazy, how can we see any assets? Well, for one, in dealing with families that have defects of sight, I would point out to them that, in some cases, a worse handicap is loss of hearing, which in turn produces a greater effect in the intellectual and linguistic retardation of the child. There are many more blind graduates from college than there are college graduates with defects of hearing. We can reframe many of these liabilities within the context of assets. I know it sounds crazy, it sounds paradoxical, but I am willing to defend that position. There are assets in the system. As it can be shown, even the child that has severe defects of the face and the head, has a complete body, and can grow into a fairly articulate human being. I like to insist that we consider the possibility of positive reframing. Positive reframing of liabilities into assets. Positive reframing in terms of comparison with worse happenings.

Finally, we need to pay attention to the discounting of feelings. Do not allow that to happen. Allow for feelings to be dealt with. Make sure that no one is discounted if they cry or feel bad. In fact, those are the individuals to consider as models for the rest of the family. Allow for the mourning process to go on until it is essentially out of the family system. After this mourning is out of the system, then you can consider alternative courses of action, rational or irrational.

The dynamite that we are dealing with is hurt feelings. Hurt feelings can unite us and hurt feelings can separate us. It is up to us to deal with them in the most constructive fashion, including the most constructive one, which is to do absolute nothing but to recognize them and express them openly without reproach.